CLAIMS

1. A nucleic acid molecule comprising at least one fragment of the human *FACL4* gene that encodes for a functional portion of the FACL4 protein to be used in the diagnosis of MR-associated syndromes.

2. A nucleic acid molecule comprising at least one fragment of the human *FACL4* gene that encodes for a functional portion of the FACL4 protein to be used in the therapy of MR-associated syndromes.

3. A method to detect in a subject at least one mutation of the gene encoding for the human FACL4 protein, located on the X chromosome, comprising the phases of:

a) collecting a specimen containing a sufficient quantity of the subject's DNA or able to be reproduced in culture;

b) isolating the DNA from the sample;

c) exponentially amplifying the DNA using as primer pair for the amplification reaction at least two oligonucleotides able to amplify a fragment of the human *FACL4* gene, in which the fragment encodes for a functional portion of FACL4 protein;

d) detecting in at least one amplified fragment any mutations compared with a healthy control.

4. A method according to claim 3 in which the exponential DNA amplification phase is performed using primer pairs for the amplification reaction able to amplify the entire coding portion of the human *FACL4* gene.

5. A method according to claim 4 in which the exponential DNA amplification phase to amplify the entire coding portion of the human *FACL4* gene will comprise the use of the following primer pairs:

Exon 3:	5' GTGAGCACATTTAGCTTAAG 3',
	5' ATCAATTGTGCTATCAACTTG 3';
Exons 3 and 4:	5' CTTCTTCAGCACAATAAGGC 3',
	5' GCATACTTAAAACGCACTCG 3';
Exon 5:	5' CCGCTCATAGCTTCTGTATG 3',

5' AACAATTCTCACATGCAAGC 3';

Exons 6 and 7: 5' AGACTGACTTCAATAATATCC 3',
5' TCATTTGTTTCCCTAACCTAC 3';

Exon 8: 5' ATTGATAGCTTATCGTTATGC 3',
5' AATGCTGAACATGAACTCTG 3';

Exon 9: 5' ATGATAAAGCTCTTGGTATTTC 3',
5' TGCAGCATCATACGATCATG 3';

Exon 10: 5' AATTCCAAGTGTAACTTCTG 3',
5' TAAAAGGTCCAAGTACGATC 3';

Exon 11: 5' ACTGTCTCCATTCCTTTCAG 3',
5' ACCTTATGATCATGGTGGTG 3';

Exon 12: 5' GAGGAATCTTTCCCAGAGC 3',
5' ATTAGTAGCAGCTGATACAG 3';

Exon 13: 5' TATTCCCAGTGCATTGGTAC 3',
5' GAAAGTCATAAAGCTGACAG 3';

Exon 14: 5' CTAATGTTCTCTCATAAAGTG 3',
5' GAACTAATGGAACCATCAAC 3';

Exon 15: 5' CAGTCAGAATTGCATATACC 3',
5' AAGAGAAGACTATGTTACCC 3';

Exon 16: 5' TTGGAATTATCTGTACTGTAC 3',
5' AGCCTAATGCAAAAGACATC 3';

Exon 17: 5' ACTCCTTTCTCGTCTCTTTC 3',
5' TAGAGGTTGAAAACCACCAG 3'.

6. A method according to claims 3 to 5 in which the phase of demonstrating, in at least one amplified fragment, mutations compared with a healthy control will be done by direct sequencing or the SSCP method.

7. A diagnostic kit for MR-associated syndromes, using the method according to claims 3 to 6, comprising:

a) at least one pair of primer oligonucleotides for the exponential amplification reaction of at least one fragment of the human *FACL4* gene,

in which the fragment encodes for a functional portion of the FACL4 protein;

b) a control DNA from a subject not affected by XLMR.

8. A kit according to claim 7 in which the oligonucleotide primer pairs for the amplification reaction are able to amplify the entire region coding for the *FACL4* gene.

9. FACL4 protein or a functional portion thereof for the diagnosis of MR-associated syndromes.

10. FACL4 protein or a functional portion thereof for the therapy of MR-associated syndromes.

11. A method for the determination of the enzymatic activity of FACL4 protein in a biologic sample, comprising the phases of:

a) collecting a biological sample from the subject, in which the sample is comprised in the group of biological fluids, lysed lymphoblastoid cells, leukocytes;

b) incubating the sample in an appropriate reaction mixture containing arachidonic acid;

c) detecting arachidonyl-CoA production.

12. A method according to claim 11 in which the detection of arachidonyl-CoA is performed using labeled arachidonic acid.

13. A diagnostic kit for MR-associated syndromes to work the method according to claims 11 or 12, comprising:

a) Lysis buffer, with appropriate protease inhibitors and/or reduction agents;

b) Coenzyme agent A and Adenosine 5'triphosphate (ATP);

c) Cold arachidonic acid and ^{14}C-labeled arachidonic acid.